AGENDA

1. Welcome and introduction from participants
2. Housekeeping and ground-rules
3. TREAT-NMD and TGDOC overview
4. Introduction to TREAT-NMD LGMD Global Registry and Core dataset development experiences in other diseases
5. Q&A
6. Break
7. Stakeholders presentations (Patients' representative, clinicians and registry curators, and pharmaceutical companies)
8. Breakout room discussions (mixed groups)
9. Feedback from groups
10. Curated questions sessions from group sessions
11. Break
12. Introduction to Meeting 2 and 3
13. Close and thank you
HOUSEKEEPING AND GROUND RULES

1. This meeting is being recorded.
2. You may have your camera on or off.
3. Please ensure that your microphone is muted at all times.
4. If you experience any connectivity issues and your camera is on, please turn it off as this may improve the situation.
5. During the presentations please write any comments or questions in the chat.
6. During the facilitated discussion/question and answer sessions if you wish to ask a question please raise your hand and the facilitator or host will invite you to unmute yourself.
7. In the breakout session please be prepared to offer your views and opinions and listen to the views and opinions of others.
TREAT-NMD AND TGDOC INTRODUCTION

LGMD CORE DATASET PROJECT
1ST MEETING
17th September 2020

Ben Watling – CEO TREAT-NMD Enterprise
Anna Ambrosini – Incoming TGDOC Chair
What is TREAT-NMD?

Advancing diagnosis, care and treatment for those living with neuromuscular diseases around the world...

- A Global Membership Network for the rare neuromuscular field, providing an infrastructure for ‘collaboration’ to ensure that the most promising new therapies reach patients as quickly as possible.

- Launched in 2007, we have focussed on the development of networks and tools that industry, clinicians and scientists need, in order to bring novel therapeutic approaches through preclinical development and into the clinic, and on establishing best-practice care for neuromuscular patients worldwide.
What is TREAT-NMD?

For Patients
- Promising research results are starting to be translated into treatments
- Work on standardized care guidelines aims to improve numbers of patients receiving optimal care

For Industry
- Supporting clinical trial readiness by providing disease specific natural history data from our associated Member Registries
- Accessing the appropriate patient cohorts for clinical trial planning and recruitment

For clinicians & researchers
- Working on support tools such as validated clinical outcome measures and standard operating procedures for research protocols to aid therapeutic development

TREAT-NMD addresses numerous issues, uniting the stakeholders in the community and providing an infrastructure that is accelerating research and therapy development, increasing collaboration, improving patient care and helping to support ‘clinical trial readiness’ on an international scale.
Who are the Executive Committee?

- Chair - Annemieke Aartsma-Rus
- Interim Vice-Chair – James Dowling

- Fabiola Bertinotti
- Marie-Christine Ouillade
- Volker Straub
- Andoni Urtizberea
- Elizabeth Vroom

- Annamaria De Luca
- Gillian Butler-Browne
- James Dowling
- Linda P Lowes
- Patient Representative TBC
TREAT-NMD Structure

TEC
- Masterclasses
- Standards of Care
- Family Guides

TREAT-NMD Executive Committee

Disease Specific Advisory Boards (KOLs)

TREAT-NMD Enterprise Board
- Operational
- Procedural
- Legal/Contracting
- Financial

General
- Membership Enquiries
- TREAT-NMD Conference
- Network Enquiries

TGDOC
- Registry Membership & Affiliation
- Registry Data Enquiries
- (Academic Research, Trial Feasibility & Recruitment)

TACT
- Clinical Trial Planning, Development, Protocol Advice
- Panel Review
TREAT-NMD
Global Data systems Oversight Committee

(TGDOC)
Composition and role

Responsible for reviewing the requests for data from the TREAT-NMD Global Registries Network

- Established in 2007
- Chaired by a troika
- Annual face to face meeting
- 79 affiliated registries

TGDOC

TREAT-NMD registry curators

Patients organizations

TREAT-NMD alliance representatives
TGDOC Chairs

Anna Ambrosini
Incoming chair

Craig Campbell
Current chair

Nathalie Goemans
Outgoing chair
TGDOC Structure
Core Members are registries that participate in global enquiries (collecting core dataset(s), updated regularly, CDA in place, correct consent, etc.).

Affiliated members do not participate in global enquiries, but are otherwise engaged with TGDOC. Can take part in internal surveys & dataset projects. Includes international ultra-rare disease-specific registries (with own enquiry processes) and registries working towards TGDOC affiliation.

Wider Registries Community includes all active patient registries we are aware of, who do not participate directly in any TGDOC projects. We list them on website & encourage them to join in dataset creation/expansion projects.
Aims: usually to establish where patients are located for clinical trial feasibility / recruitment

- Patient numbers can be stratified by any dataset items
- Can request data from all disease-specific registries, or just for certain countries

Only Active, TGDOC Core member registries can participate in formal enquiries

- Must be collecting Core Minimum Dataset for specified disease area(s)
- Must have patients’ consent to share data
- Must have Confidential Disclosure Agreement in place
- Patient data must be updated at least annually
- Patients must have a genetically confirmed diagnosis
Global Data Enquiry Process

Enquiry received from industry or academia

Global Registry Network

National registry

National registry

National registry

National registry

National registry

National registry

TGDOC Governance

TREAT-NMD requests Data

National registry returns Data
# New TGDOC Membership Criteria

<table>
<thead>
<tr>
<th>Affiliation Criteria</th>
<th>Core Member Registry</th>
<th>Affiliated Member Registry</th>
<th>Unaffiliated Registry</th>
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<tbody>
<tr>
<td>Disease subgroups</td>
<td>DMD / SMA / DM1 / FSHD/ <strong>LGMD</strong></td>
<td>Any / all NMDs</td>
<td>Any / all NMDs</td>
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<tr>
<td>Allow registry info on TREAT-NMD website</td>
<td>Essential</td>
<td>Essential</td>
<td>Strongly Preferred</td>
</tr>
<tr>
<td>Abide by Charters &amp; data protection regulations</td>
<td>Essential</td>
<td>Essential</td>
<td>Strongly Preferred</td>
</tr>
<tr>
<td>Must complete annual Registries Review Survey</td>
<td>Essential</td>
<td>Essential</td>
<td>Strongly Preferred</td>
</tr>
<tr>
<td>Collect all mandatory items in Core Dataset(s), and HE if possible</td>
<td>Essential</td>
<td>Strongly Preferred</td>
<td>Strongly Preferred</td>
</tr>
<tr>
<td>Participate in dataset revision process</td>
<td>Essential</td>
<td>Strongly Preferred</td>
<td>Strongly Preferred</td>
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<tr>
<td>Participate in disease subgroups</td>
<td>Essential</td>
<td>Strongly Preferred</td>
<td>n/a</td>
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<tr>
<td>Attend TGDOC Annual Curators’ meeting</td>
<td>Essential</td>
<td>Strongly Preferred</td>
<td>n/a</td>
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<tr>
<td>Engage with TGDOC Publications Committee</td>
<td>Essential</td>
<td>Essential</td>
<td>n/a</td>
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<tr>
<td>CDA in place and DoI submitted</td>
<td>Essential</td>
<td>Strongly Preferred</td>
<td>n/a</td>
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<tr>
<td>Participate in Surveys / Non-Core dataset Queries</td>
<td>Strongly Preferred</td>
<td>Strongly Preferred</td>
<td>n/a</td>
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<tr>
<td>Participate in Enquiries</td>
<td>Essential</td>
<td>Only relevant for Core Members</td>
<td>Only relevant for Core Members</td>
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<tr>
<td>Abide by Voting SOP</td>
<td>Essential</td>
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Any questions?
TREAT NMD LGMD Global Registry
LGMD CORE DATASET PROJECT
1st MEETING
17th September 2020

Sonia Segovia - TREAT-NMD Project Manager
TREAT-NMD actions in LGMD field

- Educational
  - Masterclasses
- Standards of care
  - Guidelines
  - PROMs
- Registries
LGMD registries actions

**Phase A.** Development of LGMD Core Dataset and network

**Phase B.** Roll out of LGMD Core Dataset + development of URP

**Phase C.** Development extended LGMD dataset
Why now?

- The current LGMD registry landscape is fragmented.
  - Multiple small, subtype registries
  - General neuromuscular diseases registries

- Acceleration of therapeutic developments
  - Recruitment for clinical trials
  - Post-marketing surveillance

NO CORE DATASET FOR LGMD

LGMD CORE DATASET FOR LGMD registries
Objectives. Phase A. Development of LGMD Core Dataset and network

Establish the **LGMD global registry network** as affiliates of TREAT-NMD and reach consensus on a feasible **core data set**, that all members agree to collect.
Project team

Michela Giugleri
Principal Investigator

Sonia Segovia
Project Manager

Heather Hildsen
LGMD project liaison

Ben Watling
CEO TREAT-NMD
<table>
<thead>
<tr>
<th>TREAT-NMD LGMD Advisory Board</th>
<th>Institutional Support</th>
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<tbody>
<tr>
<td>Jordi Diaz Manera</td>
<td>Newcastle University</td>
</tr>
<tr>
<td>Michela Guglieri</td>
<td>Newcastle University</td>
</tr>
<tr>
<td>Volker Straub</td>
<td>Newcastle University</td>
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<tr>
<td>Johanna Palmio</td>
<td>Tampere, Finland</td>
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<tr>
<td>Linda Lowes</td>
<td>Children's Nationwide Columbus Ohio</td>
</tr>
<tr>
<td>Craig Campbell</td>
<td>University Western Ontario</td>
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<tr>
<td>Nathalie Goemans</td>
<td>Leuven University</td>
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<tr>
<td>Andoni Urtizberea</td>
<td>Hendaye</td>
</tr>
<tr>
<td>Maggie Walter</td>
<td>Munich University</td>
</tr>
<tr>
<td>Nick Johnson</td>
<td>VCU Richmond USA</td>
</tr>
<tr>
<td>Elena Pegoraro</td>
<td>University of Padova</td>
</tr>
<tr>
<td>Tanja Stojkovic</td>
<td>Institute of Myology, Paris</td>
</tr>
<tr>
<td>Anna Mayhew</td>
<td>Newcastle University</td>
</tr>
<tr>
<td>Edmar Zantoneli</td>
<td>Sao Paolo, Brazil</td>
</tr>
<tr>
<td>John Vissing</td>
<td>Rigshopitalet, Copenhagen</td>
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<tr>
<td>Laura Rufibach</td>
<td>Jain Foundation</td>
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<tr>
<td>Jenn Levy</td>
<td>Coalition Cure Calpain 3</td>
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<tr>
<td>Jordi Diaz-Manera</td>
<td>Newcastle University</td>
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**TGDOC chair**

- Craig Campbell
- Nathalie Goemans
- Anna Ambrosini
## Working group

<table>
<thead>
<tr>
<th>Physician/ Registry curators</th>
<th>Institution/registry</th>
</tr>
</thead>
<tbody>
<tr>
<td>John Vissing</td>
<td>Copenhagen Neuromuscular Center</td>
</tr>
<tr>
<td>Marlene Jagut</td>
<td>Belgian Neuromuscular Disease Registry</td>
</tr>
<tr>
<td>Maggie Walker</td>
<td>Friedrich-Baur-Institute, Dept. of Neurology, Klinikum der Universität Munich, Germany</td>
</tr>
<tr>
<td>Meredith James</td>
<td>John Walton Muscular Center</td>
</tr>
<tr>
<td>Erin o'Farrell</td>
<td>Canadian Neuromuscular Disease Registry</td>
</tr>
<tr>
<td>Monique Ryan</td>
<td>Australian Neuromuscular Disease Registry</td>
</tr>
<tr>
<td>Giacomo Comi</td>
<td>Telethon</td>
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<tr>
<td>Lindsay Alfano</td>
<td>Nationwide Children Hospital</td>
</tr>
<tr>
<td>Rasha el Sharif</td>
<td>Egypt Neuromuscular Disease Registry</td>
</tr>
<tr>
<td>Miriam Rodrigues</td>
<td>New Zealand Neuromuscular Disease Registry</td>
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<tr>
<td>Francesc Pla</td>
<td>Spanish Neuromuscular Diseases Registry</td>
</tr>
<tr>
<td>Damjan Osredkar</td>
<td>University Medical Center Ljubljana, Children's Hospital Ljubljana</td>
</tr>
<tr>
<td>Rachel Alvarez</td>
<td>Cure CMD-Congenital Muscle Disease International Registry</td>
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<table>
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<th>Patient representatives</th>
<th>Institution/registry</th>
</tr>
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<tbody>
<tr>
<td>William Lowery</td>
<td>LGMD-1D DNAJB6 Foundation</td>
</tr>
<tr>
<td>Ralph Yaniz</td>
<td>LGMD2L Foundation</td>
</tr>
<tr>
<td>Mandine Casado</td>
<td>AFM</td>
</tr>
<tr>
<td>Monica Suarez</td>
<td>ASEM</td>
</tr>
<tr>
<td>Stefania Pedroni</td>
<td>UILDM Unione Italiana Lotta alla Distrofia Muscolare</td>
</tr>
<tr>
<td>Sarah Shira Emmons</td>
<td>Jain Foundation</td>
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<tr>
<td>Jennifer Levy</td>
<td>Coalition to cure calpain 3</td>
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<tr>
<td>Lindsay Murphy</td>
<td>Global FKRP registry</td>
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<th>Pharmaceutical companies</th>
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<tr>
<td>Sarepta</td>
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<tr>
<td>Affinia</td>
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<tr>
<td>AskBio</td>
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<td>Catabasis</td>
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<td>Edgewise</td>
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Working Plan. Phase A. Development of LGMD Core Dataset and network

Meeting 1
- Registry objectives

Meeting 2
- Data selection

Meeting 3
- Data feasibility assessment

Meeting 4
- Core dataset
- Data dictionary
- Implementation Plan

Core Dataset development

March 2020
17th September 2020
November 2020
January 2021
March 2021

All stakeholders
Clinicians/ registry curators/patients' representatives
All stakeholders
Meeting 1. LGMD registries objectives definition

**MAIN OBJECTIVES OF THE REGISTRY**

- **Objectives survey**
- **Results compilation**
- **Clinicians/registry curators**
- **Patients representatives**
- **Pharmaceutical companies**
- **Pre-meeting calls**

**Process Flow**

1. Objectives survey → Results compilation → Pre-meeting calls
2. Meeting 1 → TREAT-NMD and TGDOC introduction → Presentation of survey results → Discussion (Facilitator)
Meeting 2 and 3. Core Dataset development

- Meeting 2: Data selection
- Meeting 3: Assessment of feasibility
Meeting 4. Review of Core Dataset and implementation plan

- Core Dataset
- Data dictionary
- Implementation plan
Challenges of the project

• Different realities within the registries

• Different stakeholders needs and expectations

• Different diseases
Phase B. Roll out of LGMD Core Dataset + development of URP

**TREAT-NMD Enterprise. Universal Registry Platform (URP)**

- Project kicked off on 12	extsuperscript{th} August 2020

- First system build for Clinician Portal is currently underway for DMD and SMA datasets

- Will soon have access to the system for alpha testing of ‘Sprint 1’

- Thrive – graphic design company engaged
Phase B. Roll out of LGMD Core Dataset + development of URP

**TREAT-NMD Enterprise Universal Registry Platform (URP)**

- Invitation to registries to assist with alpha testing for upcoming Sprints through to Beta testing in October for Clinician Portal Testing
- Patient Portal development to begin 4th November 2020
- Central Data Warehouse (CDW) build will ensure the safe management of data input into the system - whether directly or from a registries own system
OTHER EXPERIENCES IN CORE DATASET DEVELOPMENTS
SMA AND DMD
Original Core Dataset (v0)  Expanded Core Dataset (v1)

Original Mandatory Items
- Demographics
- Best & current motor function
- SMA type
- SMN2 Copies
- Genetic test result
- Wheelchair use
- Clinical trials
- Pulmonary function
- Feeding function
- Clinical diagnosis
- Scoliosis surgery
- FVC results if done
- Hospitalisations & co-morbidities
- PRO: Clinical/Total Global Impression
- Family history

Original Highly Encouraged Items
- Participation in other registries

Expanded Mandatory Items
- Enrolment & consent
- Genetic diagnosis
- SMA type & onset age
- Best & current motor function extended
- Medications & disease-modifying therapies
- Hospitalisations & co-morbidities
- PRO: Clinical/Total Global Impression
- Demographics incl. PPRL fields
- TGI according to clinician
- Screening programme & method of testing
- Electrophysiology & biomarkers taken (Y/N)
- Date & cause of death
- Clinical observations incl. contractures

Expanded Highly Encouraged Items
- DOB, Sex, Country
- FVC results if done
- Wheelchair use
- Feeding function
- IV & NIV use
- Allopathic drugs
- Clinical trial participation
- Clinical observations incl. contractures
- Participation in other registries or NH studies

131 items  ➔  85 mandatory  ➔  37 ‘parent’ mandatory
DMD Dataset Development

1. The original DMD dataset was developed by expert consensus in 2007-2008
2. Its development was funded as part of the original TREAT-NMD FP6 EU project
3. The DMD dataset was launched in 2008 and revised in 2013
4. A working group of key DMD opinion leaders were again consulted in 2019 on a new expanded core dataset
5. The new expanded core dataset is being evaluated by 13 pilot registries from Europe & North America
6. The pilot runs from July to October 2020, whereupon its findings will be reviewed by the working Group.
Thank You
LGMD CORE DATASET PROJECT
1ST MEETING
17th September 2020

Introduction to Meeting 2 and 3 – identifying the core data set
Previous task
MEETING 2. Data selection

Purpose

- To define the core data set in line with the registry’s objectives
- To ensure that all stakeholder views are taking into account when defining the core data set
- To establish a priority order of data points
MEETING 2. Data selection

Process

• Interactive Zoom session-Breakout rooms

• The snowballing technique – building a consensus
  • Paired discussions – small group discussions – discussions in plenary

• Facilitated process

• 2 hour meeting
MEETING 2. Data selection

Building a consensus

**MoSCoW** prioritisation technique
- Must have
- Should have
- Could have
- Won’t have

Individual views – small group consensus – large group (plenary) consensus
MEETING 3. Feasibility

Purpose

- To determine the feasibility of collection and the value of each data set

- To ensure that there is consensus of core data set and shared views about feasibility and value.
MEETING 3. Feasibility

Process

- Follows on shortly after meeting 2 (Defining the core data set)
- Interactive Zoom session
- Using Google Jamboard
  - Interactive sorting exercise in groups with data sets
VE – very easy
E – easy
VD – very difficult

Feasibility

Low
Value
High
MEETING 3. Feasibility

Jamboard

• Classification of data
• Feasibility of collection/value of data

• Start with Must Have and Should Have from meeting 2
MEETING 3. Feasibility

Outcomes

- Agreed core data set
- Ranked by feasibility and value
ANY QUESTIONS?